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ABSTRACT

Abstracts of 13 papers given at a 1983 conference on autistic children are presented: "Autism--Is There a Neurological Basis?" (G. McKhann); "The Syndrome of Autism: A Medical Model" (E. Ritvo) (full paper); "Neuropsychological Testing of Peripheral and Central Communicative Disorders" (H. Mark); "A Psychiatrist Views the Neurological Basis of the Communicative Disorders" (P. Wolff); "Behavioral Mannerisms of the Blind Infant and Child (A Synopsis)" (J. Bond); "Specific Learning Disability: A Communicative Disorder" (B. Shapiro); "Normal and Abnormal Development of Feeding (J. Bosma); "The Relationship between Oromotor Dysfunction and General Movement Development" (R. Alexander); "Oromotor Facilitation in Pre-Speech" (R. Alexander); "Management of the Child with Speech Motor Dysfunction" (S. Blackstone); "The Neurology of Oromotor Dysfunction and Language Disorders" (H. Singer); "Genetics of Autism" (J. Farber); and "Metabolic Approaches to Autism and Related Disorders" (H. Moser). (CL)

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[Autistic Children Developmental Disabilities Conference Abstracts of Papers Presented]

Johns Hopkins University

Baltimore, Maryland, March 14-16, 1983

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AUTISM: IS THERE A NEUROLOGICAL BASIS?

Guy M. McKhann, M.D.

My discussion will review some of the data suggesting that autism has a neurological basis. An attempt will be made to try and relate the symptoms expressed by children with autism with possible localizations within brain. Finally, I would like to suggest some directions for future research on the neurological basis of autism.

The Syndrome of Autism: A Medical Model

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This manuscript was prepared at the request of Alfred M. Freedman, M.D., Editor-in-Chief, Integrative Psychiatry.

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I wish to thank all my collaborators at U.C.L.A. and other medical centers throughout the world. It has been a privilege to work with them over the past 20 years and to have shared the excitement of unraveling a few of the many mysteries surrounding autism. Equal thanks go to our patients, their families, control subjects, and volunteer workers. Without their cooperation we would still be back on ground zero, unable to conduct our first experiment.

The writing of this manuscript and much of my recent research has been directly supported by the generosity of the Max and Lottie Dresher Fund and the Bennin Fund. Federal grants currently supporting my research include: MH-29248, MH-30897, MH-31274, MCHP 922, and the V. A. Medical Research Service. Finally, invaluable assistance was provided by my secretarial co-worker, Bernice Heyert, and my chief research associate, Anne Mason Brothers.

Abstract

Child psychiatrists throughout the world recently agreed to consider autism a behaviorally defined syndrome. To establish the diagnosis symptoms must be present before 30 months of age (to distinguish it from later onset diseases and syndromes) and include characteristic disturbances of (1) developmental rates and sequences, (2) responses to sensory stimuli, (3) speech, language, and gestures, and (4) the capacity to relate to people, objects, and events.

As a result of this consensus scientists from many disciplines are now able to apply sophisticated research techniques to groups of uniformly diagnosed patients. This exciting work is best viewed from the perspective of the medical model which relates symptoms to underlying pathologic processes, seeks etiologies and, finally, leads via deduction to rational (specific) therapies.

Breakthroughs are at hand. Subtypes of autism based on neuropathology and etiology are emerging. We are now designing and testing therapies aimed at mitigating specific pathologic processes. Possibilities for prevention are just over the horizon. We are fortunate to be practicing at this historical moment in the history of medicine.

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The Syndrome of Autism: A Medical Model

Only a few short years ago patients with the syndrome of autism were labeled as having childhood psychosis, severe emotional disability, atypical ego development, childhood schizophrenia, symbiotic psychosis, early (primary) or late (secondary) onset autism, or mental retardation with autistic features. Indeed, the most critical factor in evaluation was where the diagnostician had been trained, not the patient's symptoms. Therapy was just as idiocyncratic: ranging from parentectomy and psychoanalysis (exorcise and neutralize psychic toxins) to ECT and lobotomy (stun and lacerate physical toxins). Most pathetically, the same patients were often prescribed alternating courses of these widely differing "therapies" as their families floundered from clinic to clinic vainly seeking ways to help their children.

Fortunately, as we enter the 1980's, the situation has dramatically improved. Today we are on the threshold of a new era of research and understanding--an era during which we shall hopefully learn how to prevent some types of autism and discover successful treatments of other types. This optimism is based upon the fact that scientists throughout the world, following the classical medical model, are beginning to focus their attention on the problem. This model begins with an objective definition of a syndrome, then identifies pathological processes that could produce its symptoms and developmental delays, next pinpoints etiologic factors, and finally leads by deduction to the discovery of "rational therapies" (i.e., ones that eliminate

ongoing etiologic factors, normalize pathologic processes, or make up developmental lags). We are at an exciting moment in the history of medicine and a fortunate one for our autistic patients and their families.

At your Editor's request, this essay will present a brief status report on the state-of-the-art, research-wise, regarding autism. How we arrived where we are is another story. For those interested in reviewing the scientific literature that has appeared since Kanner's original 1943 description of the syndrome in his classic paper entitled "Autistic Disturbances of Affective Contact" (1), I can recommend three sources. First is a volume entitled "Autism: Diagnosis, Current Research, and Management" I edited with three colleagues at UCLA--Drs. B. J. Freeman, Edward M. Ornitz, and Peter E. Tanguay (2). It contains several review chapters and covers issues of advocacy. Second is a review article by Dr. Edward M. Ornitz and myself published in the American Journal of Psychiatry entitled "The Syndrome of Autism: A Critical Review" (3). Lastly, is a comprehensive and scholarly article entitled "Infantile Autism Reviewed: A Decade of Research," by Drs. DeMyer, Hingtgen, and Jackson, recently published in Schizophrenia Bulletin by the National Institute of Mental Health (4).

Table I contains a schematic overview of the medical model as applied to the syndrome of autism. Each box contains headings which caption specific areas of knowledge (or our lack thereof). It is offered to help organize your understanding of our current knowledge of autism and to serve, in a modified order, as a table of contents for what follows. (INSERT TABLE I)

The Clinical Syndrome

In 1978 the Professional Advisory Board of the National Society for Children and Adults with Autism formulated a "Definition of the Syndrome of

Autism" (5). It represented a consensus among professionals from different countries and disciplines and formed the basis for a similar and overlapping definition published in the Third Edition of the American Psychiatric Association's Diagnostic and Statistical Manual (DSM-III). Both definitions are summarized in the Appendix for those not familiar with the new criteria. They are based on assumptions and facts which contradict some long thought and long taught misbeliefs about the subject, and may require revision and updating of your thinking. For example, it is now agreed that:

1. Autism is a clinically (behaviorally) defined syndrome. This is because objective pathognomonic biomedical markers common to all cases have not yet been identified. Like all syndromes (for example, pneumonia, epilepsy, hypertension, diabetes) autism is assumed to be composed of many sub-types, each with different etiologies, pathologies, and possible treatments.
2. Autism begins prior to 30 months of age. This time was chosen in order to distinguish it from other developmental syndromes and diseases which begin later in childhood. Since many patients are not seen until older it means that the diagnosis is often made retrospectively, based upon careful history taking, reviewing of medical and school records, and scanning baby books and home movies, etc.
3. Autism is lifelong. While symptoms fluctuate, abate, and may even disappear, if they were present when a patient was less than 30 months of age, the diagnosis remains. Once autistic, always autistic. If and when cardinal symptoms do disappear, the diagnostic code becomes DSM-III 299.01, Autism, residual state.

4. Autism has two distinct types of clinical onset. First are patients whose developmental delays and symptoms are observable during the first few weeks and months of life. Second are patients who display normal development until 12 to 24 months of age, after which regressions and plateaus in development begin and specific symptoms first appear. It is not yet known if an association exists between type of onset and course of illness.
5. Autism is ubiquitous. It occurs in all parts of the world, in all races and colors, and in all types of families. (It is not found more often in certain socio-economic groups as is mistakenly stated in DSM-III. That notion was based on outdated research. Indeed, no social or psychological characteristics of parents or families have proven to be associated with autism.) Boys are affected more often than girls by a ratio of four or five to one and many families have more than one autistic child. Autistics have a normal life expectancy and the vast majority (over 90%) require lifelong social support systems due to their developmental handicaps and continuing symptomatology.
6. Autism frequently occurs in association with other syndromes, specific diseases, and developmental disabilities. For example, over 80 percent of all autistic patients also have the syndrome of mental retardation (i.e., score in the "mentally retarded" ranges on I.Q. testing). This is because the brain pathology which produces the symptoms of autism also effects cognitive development and symbol processing. Epilepsy, motor incoordination, Fragile X, and severe allergies are other frequent "fellow traveler" syndromes.

A final word about diagnosis. No where else do we need to be more skillful in practicing our art than when dealing with a youngster who might be autistic and concerned, vulnerable parents. We must go slowly. If not absolutely sure (for instance, when patients are less than two years old) or because symptoms and history are not clear upon first review, I simply tell the parents the three hardest words to say in any language, "I don't know." Then I arrange to have another "look see" in a few weeks or a month. Several such clinical snapshots may be necessary before I can piece together a mental movie of the child's development--a movie of sufficient detail to reveal the developmental disturbances and symptoms indicative of autism. But it is preferable to go slowly and be certain, than either miss another diagnosis or needlessly alarm a family by saying autism may be present. Autism is a "hard" diagnosis to make, just as leukemia is "hard" to diagnose because it portends such seriousness. But this only means we must not deny its presence when it is there. (How many parents of an autistic have heard from a physician when first they sought help, "Oh, he'll grow out of it."?) We must always proceed to make the diagnosis very judiciously and with all due humility and sensitivity for the feelings of parents and siblings.

Possible Etiologies

At present no specific cause or causes have been identified. Importantly though, we do know that autism cannot be produced by psychological trauma, physical abuse, or bad parenting as was once speculated. Such traumatized infants are very peculiar and have very deviant development, but their developmental abnormalities and symptoms are far from those diagnostic of autism. Indeed, the past decades of psychotoxic theorizing are a black mark

in medical history due to the needless harm that was inflicted on families and the wasted years of patients' lives that passed in fruitless psychotherapy. Even more discouraging though, is the fact that many of our colleagues here in the United States and elsewhere still, to this day, cling to these outmoded and disproven notions, refusing to update their knowledge.

1. Genetic factors: These have been suggested as playing a part in the etiology of almost all major neuropsychiatric diseases and syndromes. Table II contains a somewhat fanciful schema which relates genotypes and phenotypes along the dimensions of "time of onset" and "degree of pervasiveness." On the top line is indicated the possibility that autism, childhood onset schizophrenia, and adult onset schizophrenia share a common genotype. Hopefully this and similar hypotheses will soon become testable by the discovery of markers for one or the other of these major syndromes. (INSERT TABLE II)

Focusing down from the wide angle view of Table II onto just autism, it is highly likely that genetic factors may play a role in causing certain cases (a genetic sub-group). These genetic influences could act directly via a recessive gene transmitting a specific defect as in P.K.U. or hemophilia. In autism such a defect could produce structural brain abnormalities, enzymatic or neurotransmitter pathology, defective brain maturation (e.g., arborization) or other types of pathology.

Indirect genetic influences may also be responsible for other subtypes of autism. For example, a genetically determined decreased resistance to viruses or other interuterine or post-natal brain insults may be the culprit. Could a slow viral encephalitis account for those patients in whom symptoms begin after an apparently normal development of 15 months or so?

To explore these hypotheses I established the UCLA Registry for Genetic Studies in Autism (6). From its inception in January 1980 to October 1, 1982, 281 families have enrolled. They include (by parental report) (a) 22 sets of monozygotic twins, all concordant for autism; (b) 18 sets of dizygotic twins, 2 sets of whom are concordant for autism; (c) 46 sets of non-twin siblings concordant for autism; (d) 6 families with 3 autistic siblings; (e) 20 families with autistic cousins; and (f) 5 parents of autistic children who have an autistic brother. These families are being intensively studied with a variety of immuno-hematologic techniques and pedigree analyses.

2. Specific viral infections have been implicated in the etiology of certain cases of autism. Strong evidence for this comes from the high incidence of autism in children known to have been infected while in utero by rubella virus. Indirect evidence supporting this same association is the observation that some autistics have decreased immunologic responsiveness to antigen challenges.

3. Traumas to the developing CNS of all types have been sought as causes of autism, but non proven--or even weakly supported by scientific data. Suggestions in the literature range from pin worms to vitamin deficiency. At this point in time it seems highly unlikely that any specific toxic agent or deficiency will be identified serendipitiously. Rather, systematic studies in the areas of neuropathology or clinical neurology may point us down a fruitful path which to date has eluded detection.

Possible Pathologic Processes

Systematic autopsy surveys of neuropathology are lacking because patients have normal longevity. The few isolated reports published to date are non-conclusive. To help remedy this situation I established in 1978 a brain bank

at U.C.L.A. in collaboration with the Professional Advisory Board of the National Society for Children and Adults with Autism, U.S.A., and other universities. To date three brains have been received, none of which revealed significant pathology upon routine autopsy. Hopefully, studies including cell density and ultrastructures on these and subsequent specimens will reveal pathognomonic types and locations of abnormalities. Areas of search are widespread as deductions from clinical symptoms indicate that widely separated brain areas may be involved. (The cortex is suggested by cognitive, speech and language, and specific symbolic processing dysfunctions; the limbic, frontal, and temporal systems are suggested by relationship and affective symptoms; the brain stem, vestibular system, and cerebellum are suggested by sleep cycle, motility and sensory response symptoms, and by decreased post-rotatory nystagmus.)

Clinical evidence of neuropathology in specific cases of autism is being reported from clinics throughout the world. The latest sophisticated monitoring techniques are being applied to uniformly diagnosed patients. These abnormal findings are of two types. First are structural (or wiring) abnormalities and, second, functional or metabolic disturbances.

1. Structural (or wiring) abnormalities. Pneumoencephalograms (PEG) and computerized tomography (CT) scans reveal abnormalities in otherwise clinically indistinguishable autistics. Pathologic findings range from cerebral atrophy and asymmetry to enlarged ventricles. These findings appear to be non-specific as to their cause and relationship to autistic symptomatology from the radiologic data to date.

2. Functional and metabolic abnormalities. EEG, Positron Emission Tomography (PET) Scans and biochemical assays also reveal subgroups of

autistics with abnormal findings.

EEG studies are the most numerous, most extensive, and most convincing. Also clinical experience easily confirms the research findings that autistics are increasingly likely, with age, to develop EEG abnormalities and seizures. Exact percentages are impossible to determine because of methodology problems inherent in long term follow-up research. However, in my clinical practice EEG abnormalities and seizures are rarely encountered in children less than six years old. However, about 10 percent of those followed to adulthood have developed EEG abnormalities and 5 percent seizures.

Positron Emission Tomography (PET) Scanning is a new technique which allows measurement of the rate of glucose metabolism in different parts of the brain. A preliminary report from the N.I.M.H. at the 1982 National Conference of NSAC-USA at Omaha, Nebraska, revealed that PET Scanning is being applied to a group of autistic adults. (At present, amount of radiation exposure and other technical factors preclude its experimental use in children.) The first results indicate that there may be significant difference in brain metabolism patterns between normals and autistics, but further work is necessary to confirm these clues. PET Scanning holds promise not only of providing diagnostic information, but of opening the door to therapies based upon improving brain metabolism by administering specific medications or metabolites.

Lastly, in the category of metabolic studies, serotonin (5 hydroxy-tryptophane--5 HT) must be mentioned. This cerebral neurotransmitter is also found in blood platelets where it is easily measured. Normal infants have high blood levels which decrease throughout childhood and stabilize in adulthood at lower levels. A subgroup of autistics (probably about one-third

to 40 percent) fail to show the expected age-related decrease (possibly a maturational failure?) and remain hyperserotonemic throughout life. Whether this peripheral blood finding is related to their being autistic or is just an adventitious observation remains to be determined. Knowledge of how blood serotonin levels are modulated and their relationship to brain and intersynaptic serotonin metabolism is scanty. However, this area of research has important implications for explaining clinical phenomena and therapeutics. For example, variability of specific symptomatology and developmental spurts and regressions are hallmarks of autism. Could these changes be due to alterations in neurotransmitter metabolism? Could we produce symptomatic remissions and induce developmental spurts by correcting neurotransmitter metabolism with medications, as we can in Parkinson's Disease?

Possible Treatments

At present the most we can offer our patients are "symptomatic" therapies aimed at helping them maximize their existing potentials and minimizing their neuropathologically induced handicaps. These techniques fall within the professional realms of psychology and special education, and are often augmented by behavioral control with psychotropic medication. These approaches will be discussed before venturing into the unchartered waters of prevention and specific psychopharmacotherapy.

1. Structured educational programs. Each autistic patient deserves to have his behavioral repertoire objectively assessed and to be systematically taught higher level skills. To accomplish this a field within special education called behavioral technology and based on principles of operant conditioning has evolved over the past decade. Special education teachers have been trained to apply this technique to autistics in the least restrictive

environment a patient (pupil) can tolerate. Constant objective monitoring of such educational programs, their goals and techniques is required and constant updating and changing of curricula is to be expected. While maximum gains are achieved by starting teaching at as young an age as possible, improvements have been observed up through teen years and in rare instances adulthood (one patient began speaking at age 18 years, for example). Fortunately, most school districts in the United States provide special classes or teachers for autistics and parental participation has been shown to be instrumental in achieving maximum results. In the best programs parents are regarded as co-teachers or co-therapists and assist in curriculum development, implementation, and monitoring.

2. Symptomatic control with psychotropic medication. Often times behaviors such as assaultiveness, self-destructiveness, and unmanageable hyperactivity require the temporary use of "chemical straight jackets." When employed for these purposes the following guidelines should always be followed before and during therapy: (a) Target behavior should be specified in writing and baseline frequency counts obtained. (b) Dosages should be started at low levels and gradually increased until specific improvements in target behaviors occur or adverse side effects limit increases. (c) Target behaviors should be constantly monitored while medication is administered to document continuing therapeutic effects. (d) Medications should be periodically decreased and discontinued to insure that they are still required by the reoccurrence of the serious target behaviors for which they were initially instituted. Three months is generally the maximum time before instituting such "drug holidays." (e) Extensive clinical experience reveals that symptoms may recur while a patient is on the optimal dosage of a psychotropic medication. Often, improvement can be reinstated by simply substituting one psychotropic

with another.

3. Specific neuropharmacologic treatment. My search for effective medications began over a decade ago, sparked by two puzzling observations. First, patients were having unpredictable spurts, regressions, and changes in symptoms. (Such variability has since become one of the diagnostic hallmarks of autism.) Second, parents reported rarely, but too often to be ignored, that their children improved when feverish ("calmer, uses more words, flaps and spins less, much more tuned in, looks at you"). Were these clues that the underlying pathologic process might be dynamic or metabolic? If so, this could lead to the finding of effective medications, an impossible quest if only static or structural (wiring) pathology were at fault.

At this point in time, the late 1960's, a new scientific field, neurobiochemistry, was emerging. Serotonin (5 HT) was identified as a major neurotransmitter in the brain. Initial blood studies in autistics were conflicting and confusing, with reports published of both high and low blood levels. Despite these contradictions, I wondered, could this new field of neurobiochemistry contain explanations for the mysterious changes observed in my autistic patients?

In order to anser this question I began a collaboration with Drs. Edward Geller and Arthur Yuwiler, neurobiochemists at the Brentwood Veterans Administration-U.C.L.A. Hospital. First we perfected a reliable blood assay technique to avoid prior technical problems, then developed normative data, and, finally, tested dozens of autistics. Results revealed that in normals serotonin levels are high in infancy, drop throughout childhood, and stabilize at low adult levels by the teen years. Approximately one-third of autistics failed to show this expected maturational decrease and remained hyperserotonemic throughout childhood.

We had now reached the exciting part where we could construct testable hypotheses. Do high levels of blood serotonin reflect abnormal serotonin metabolism in the brain? Are high levels of blood serotonin associated with historical factors, types of onset, developmental disturbances, I.Q., etc? Most important, could we find a medicine to lower blood serotonin, alter (improve) brain metabolism, ameliorate symptoms, and induce developmental spurts?

After months of work resulted in only approved but not funded grants (autism was a low priority item), we were left with only one choice--to attempt a simple clinical trial. We selected L-DOPA since it lowered blood serotonin as a side effect in the treatment of Parkinson's Disease. Four hospitalized hyperserotonemic autistic boys were given this medication for six months. Their blood levels fell but unfortunately they did not improve clinically.

Next, Dr. B. J. Freeman, a psychologist who had developed the Behavior Observation System for Autism and an expert on clinical and psychometric assessment, joined our team.

Armed with improved laboratory and objective clinical measures, we decided to administer fenfluramine. We chose this anorexogenic medication as it is known to lower blood and serotonin in rats and monkeys and to readily cross the blood brain barrier. A pilot study was begun by administering increasing doses to one hospitalized hyperserotonemic autistic boy. His blood level fell rapidly and remained within the normal range for two weeks. Our objective clinical measures documented behavioral improvements at the end of this brief time.

Encouraged by this result we arranged to hospitalize two hyperserotonemic boys for a six month double blind placebo crossover study (ABA type). Both

boys showed obvious clinical and significant objective improvements while on the medication, and a return of symptoms when it was discontinued. They both improved their performances on I.Q. tests (27% and 66% respectively). One patient had an identical autistic twin. He was not hospitalized or treated and showed no changes on serial I.Q. testing or clinical observations over the same time frame.

To confirm and expand these encouraging results a multicenter clinical trial of fenfluramine is now under way at several medical school throughout the United States. Hopefully, within two to three years we shall have answers to such questions as which symptoms are ameliorated by fenfluramine and in what types of patients.

Most importantly, the work with fenfluramine to date is expanding our professional horizons in child psychiatry. We are now collaborating to study brain-behavior relationships at the molecular level and in concert with rigorous objective clinical measures. What a wonderful way to begin the decade of the eighties!

4. Prevention via genetic counseling or amniocentesis and therapeutic abortion. An earlier section on etiology contains a discussion of current evidence that subtypes of autism may be caused by genetic factors. If pedigree analysis identifies specific modes of inheritance, genetic counseling will become a reality. If pathognomonic biological markers for autism are identified, individual case prevention via therapeutic abortion will become a reality. Meanwhile, we shall continue to enlarge the U.C.L.A. Registry for Genetic Studies of Autism with uniformly diagnosed multiple incidence families while research continues.

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TABLE I

A MEDICAL MODEL OF AUTISM

POSSIBLE ETIOLOGIES	POSSIBLE PATHOLOGIC PROCESSES	THE CLINICAL SYNDROME AUTISM	POSSIBLE TREATMENTS
1. Genetic Direct Indirect	1. Structural	Disturbances of: (prior to 30 mos.)	1. Structured, behaviorally based individualized education.
2. Viral Immediate Slow Indirect	2. Functional Metabolic Neurotransmitters (hyperserotonemic sub-group)	1. Developmental rates and sequences	2. Symptomatic control with psychotropics
3. Trauma to CNS Chemical Metabolic		2. Responses to sensory input	3. Specific neuropharmacologic treatment--to normalize neurotransmitters (serotonin) or other metabolism
		3. Communications (speech, language and non-verbal communication)	4. Prevention via genetic counseling or amniocentesis and therapeutic abortion
		4. Relationships to people, objects, and events	

TABLE II

A HYPOTHETICAL GENETICALLY PROGRAMMED NEUROPATHOPHYSIOLOGY

COMMON TO SERIOUS NEUROPSYCHIATRIC DISORDERS

GENOTYPE	Early Onset	PHENOTYPES	Late Onset
Most Pervasive	→ AUTISM (299)*	SCHIZOPHRENIA (295)	PARANOID STATES (297)
	Onset prior to 30 mos. of age	Onset in childhood, adolescence, adult onset	Later adult onset
	→ DEPRESSION	AFFECTIVE DISORDERS (296)	
	Onset in childhood	Manic-depressive, Bi-polar	
		Depressive, Mono-polar	
	→ ATTENTIONAL DEFICIT DISORDERS (314)		
	± Hyperactivity		
	→ SPECIFIC DEVELOPMENTAL DISORDERS (315)		
	Reading, math, language, articulation		
	→ EPISODIC IMPULSE CONTROL DISORDER (312.34)		
	(Aggressive outbursts)		
	→ STEREOTYPED MOVEMENT DISORDERS (307)		
	(Tics, Tourettes)		
More Specific	→ CERTAIN ADDICTIONS		

*Numbers in ( ) refer to DSM III.

Appendix A

National Society for Autistic Children

Definition of the Syndrome of Autism

Essential features: Autism is a behaviorally defined syndrome. The essential features are typically manifested prior to 30 months of age and include:

(1) Disturbances of developmental rates and sequences. Normal coordination of the three developmental pathways (motor, social-adaptive, cognitive) is disrupted. Delays, arrests, and/or regressions occur among or within one or more of the pathways: (a) within the motor pathway: for example, gross motor milestones may be normal, while fine motor milestones are delayed; (b) between pathways: for example, motor milestones may be normal, while social-adaptive and cognitive are delayed; (c) arrests, delays, and regressions: for example, motor development may be normal until age 2 when walking stops; some cognitive skills may develop at expected times, while others are delayed or absent; imitative behavior and/or speech may be delayed in onset until age 3, followed by rapid acquisition to expected developmental level.

(2) Disturbances of responses to sensory stimuli. There may be generalized hyperreactivity or hyporeactivity, and alternation of these two states over periods ranging from hours to

months. For example, (a) visual symptoms: these may be close scrutiny of visual details, apparent nonuse of eye contact, staring, prolonged regarding of hands or objects, attention to changing levels of illumination; (b) auditory symptoms: these may be close attention to self-induced sounds, nonresponse, or overresponse to varying levels of sound; (c) tactile symptoms: these may be over- or underresponse to touch, pain, and temperatures, prolonged rubbing of surfaces, and sensitivity to food textures; (d) vestibular symptoms: these may be over- or underreactions to gravity stimuli, whirling without dizziness, and preoccupation with spinning objects; (e) olfactory and gustatory symptoms: these may be repetitive sniffing, specific food preferences, and licking of inedible objects; (f) proprioceptive symptoms: these may be posturing, darting-lunging movements, hand flapping, gesticulations and grimaces.

(3) Disturbances of speech, language-cognition, and nonverbal communication. Symptoms may include: (a) speech: for example, mutism, delayed onset, immature syntax and articulation, modulated but immature inflections: (b) language-cognition: for example, absent or limited symbolic capacity, specific cognitive capacities such as rote memory and visual-spatial relations intact with failure to develop the use of abstract terms, concepts, and reasoning; immediate delayed, negative echolalia with or without communicative intent; nonlogical use of concepts; neologisms; (c) nonverbal communication:

for example, absence or delayed development of appropriate gestures, dissociation of gestures from language, and failure to assign symbolic meaning to gestures.

(4) Disturbances of the capacity to relate appropriately to people, events, and objects, manifested by failure to develop appropriate responsivity to people and assignment of appropriate symbolic meaning to objects. For example, (a) people: absence, arrests and/or delays of smiling response, stranger anxiety, anticipatory response to gestures, playing "peek-a-boo," playing "patty-cake," and waving "bye-bye," reciprocal use of eye contact and facial responsivity, and appropriate reciprocal responsiveness to physical contact; failure to develop a relationship with significant caretakers or excessive reliance on caretakers. For example, caretakers may be treated indifferently, interchangeably, with only mechanical clinging, or with panic on separation. Cooperative play and friendships (usually appearing between 2 and 4) may not develop. Expected responses to adults and peers (usually appearing between 5 and 7) may develop, but are superficial, immature, and only in response to strong social cues; (b) objects: absent, arrested and/or delayed capacities to utilize objects and/or toys in an age-appropriate manner and/or to assign them symbolic and/or thematic meaning.

Objects are often used in idiosyncratic, stereotypic and/or perseverative ways. Interference with this use of objects often results in expressions of discomfort and/or panic; (c) events: there may be a particular awareness of the sequence of events and disruption of this sequence may result in expressions of discomfort and/or panic.

Appendix B

DSM-III Diagnostic Criteria for Infantile Autism

A. Onset before 30 months of age.

B. Pervasive lack of responsiveness to other people (autism).

C. Gross deficits in language development.

D. If speech is present, peculiar speech patterns such as immediate and delayed echolalia, metaphorical language, pronominal reversal.

E. Bizarre responses to various aspects of the environment, e.g., resistance to change, peculiar interest in or attachments to animate or inanimate objects.

F. Absence of delusions, hallucinations, loosening of associations, and incoherence as in Schizophrenia.

299.00 Infantile Autism, Full Syndrome Present

Currently Meets the criteria for Infantile Autism.

299.01 Infantile Autism, Residual State

Diagnostic criteria for Infantile Autism, Residual State

A. Once had an illness that met the criteria for Infantile Autism.

B. The current clinical picture no longer meets the full criteria for Infantile Autism, but signs of the illness have persisted to the present, such as oddities of communication and social awkwardness.

Henry J. Mark, Sc.D.

NEUROPSYCHOLOGICAL TESTING OF PERIPHERAL AND CENTRAL COMMUNICATIVE DISORDERS

Poets and those familiar with the symptoms of autism know that many people listen to "different drummers". Of all the symptoms found in autism, none is more disturbing to the laymen and more challenging to the neuropsychologist than to find out who that drummer is and toward what goals he is leading the patient. What does the patient hear that is so competitive with what we would like him to hear or expect him to hear? What if any goals does a patient see which are so different from the goals we have for him? If and whenever his "goals" are not all that different from ours, how do we account for our frequent failures of communication? How can we explain failures to arrive at points or achieve closure in a relatively simple emotionally neutral dialogue when at other times we have successes in similarly neutral dialogues on more complex levels or issues?

Over the years we have discovered that many clues regarding the identity of the different drummers may be contained within such internally generated phenomena as the perseverations, early developmentally "imprinted rigidity", and loose language associations we so frequently see in autistic patients. We have found that very often the patient's recent experiences (frequently as recent as previous questions within a test scale) provide many clues regarding the other competing internal "different drummer". Thus, often we can

trace the internal drummer to his external origin, i.e. the internally generated phenomena to their external sources. Whenever we can, in fact, trace the different drummer to external origins, much of the mystery of the internal drummer disappears. He is in essence just another "physical event, stimulus or challenge" who basically has no volition of his own, and slavishly follows the more general laws of psychophysics. Accordingly, perseverated concepts or loosely retrieved verbal associations, word rhymes, etc. stemming from the internal drummer will compete for the patient's attention and responses with the external new words or inputs which we provide the patient in our efforts to focus the dialogue. His associations, perseverations, etc., to our words as well as our words themselves all have the value of perceived stimuli, and according to the laws of psychophysics, responses will be maintained on each perceived stimulus until it no longer changes in "Units Perceptable to the Patient".

The concept of "Units Perceptable to the Patient" is a big concept in Psychophysics as well as in Conditioning. Just recall the laws of conditioning which state that repeated unchanging (unreinforced) stimuli ultimately inhibit responses. What units of change are perceptable to the patient depends upon his personal "differential thresholds", his "JND's" within the various sensorimotor, language and association modalities and regions of the brain. The person-specific invariance of JND's, of course, has made them the basic conceptual building blocks of virtually all of sensory physiology and psychophysics and conditioning.

In brief, we are placing the heavy burden accounting for imbalances between diverging and converging thought processes, rambling conversations, behaviors which appear to lack goal directions, perseverations, contaminations, and a host of other symptoms on "location-specific" Differential Thresholds or JND's. No more mysterious distant drummers which control motivations through other laws! Psychopharmacologists as well as behavioral scientists would clearly be pleased if we could demonstrate that such a conceptual view can place observed phenomena in autism and other cognitive dysfunctions into reasonable perspectives because at least theoretically, Differential Thresholds are accessible by modern methods and techniques.

Obviously, the gaps in our scientific knowledge are enormous But organizing clinical procedures within the liberating straightjacket of the laws of parsimony and more rigorous science, not only satisfies a longing for elegance, but, as in other areas of science, can have pratical implications in technological spin-offs. Our spin-off is an improved "High Articulation Field (1)".

"What?" you say, "all this jazz about drummers and JND's just for a High Articulation Field?" Yes, and we claim no credit for the originality of the idea. It was first proposed as an essential prerequisite for progress in the Mental Health Field by Professor Cornfield, then chairman of the Johns Hopkins School of Hygiene & Public Health Biostatistics Department. The basic idea is really quite simple. Patients do not come labeled with special talents, defects, handicaps, pseudo talents (such as apparently using sentences when they can only use single words but parrot phrases), pseudo handicaps (e.g. apparent lack of organizing skills when the

real problem is in the motor-affector area which might be overcome electronically or mechanically), etc. Without labels, it requires a diagnostic search to find all of these relevant cognitive factors. It helps to know the boundaries of the field to be searched, and how these factors may be related (i.e. how they might articulate with one another in that field). In fact, without such knowledge the search pattern is likely to be chaotic, or possibly miss the most important potential talents or significant defects because they are in a hidden corner of the field or outside the field; at best, the search will be non-systematic, non-comprehensive and inefficient.

The presentation will focus on the boundaries of the field of cognitive disorders surveyed by neuropsychological testing of peripheral and central communication disorders, and demonstrate a systematic and comprehensive computer guided search for the cognitive factors mentioned which lie within this field. After describing such a computer guided diagnostic data acquisition process, we will discuss a computerized data processing system which can report and translate the diagnostic patterns found into management and treatment recommendations. The Data Base Management System currently being developed with support from our Kennedy Pediatric Rehab Unit will be able to track the natural history of cognitive syndromes and disorders both in patients with developmental disabilities and those recovering from trauma, head injury or other CNS insults. The system will ultimately provide leads on the efficacy of various cognitive diagnosis-specific management strategies and interventions selected by various services or

clinicians who share their data on these patient populations with us in a data pool. That would serve to take some of the mysteries not only out of the different drummers in autism but more generally out of diagnoses and treatments in the Mental Health field.

MARK

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A PSYCHIATRIST VIEWS THE NEUROLOGICAL BASIS OF THE COMMUNICATIVE DISORDERS

Peter H. Wolff, M.D.

The proposed discussion will not address the issue of autism; nor does it deal specifically with clinical aspects of communicative disorders by conventional definition; nor will it be presented by a practicing psychiatrist.

Instead, I propose to explore neuropsychological issues of language acquisition which presumably are critical features in most communicative disorders from an idiosyncratic perspective of motor development. Starting with Lashley's classic paper on serial order, as well as from experimental, clinical, and theoretical accounts concerning hemispheric specialization of neurological processes that control motor and language performance, I will argue that cerebral specialization for language is derivative of its specialization for particular features of sensorimotor control such as serial ordering, rapid transitions, inhibition of excessive or inappropriate movement. The concept of motor control will be used as a metaphor which refers to central programs which regulate action rather than to the skeletal muscular movements themselves. The burden of the argument will be that the cerebral hemispheres are not specialized for the control of verbal skills, cognitive processes, or any other psychologically defined category of function; but, if specialized at all, they are so for physiological parameters of function. To the extent that the left hemisphere is specialized, it is relatively more competent than the right for temporal discrimination, linear processing rates, serial reproductions, and serial ordering of output. From this perspective, it should follow that at least some children with communicative (language) disorders, and perhaps a substantial proportion will demonstrate parallel disturbances in the motor domain. However, the manifest motor disabilities must be analyzed in terms of physiologically plausible categories and the finer details of component functions in order to identify the

underlying neuropsychological deficit. In other words, gross measures of motor performance may or may not be correlated with communicative disorders, but such correlations provide no clues about the underlying mechanisms that may be shared by disorders in both domains. Finally, it will be argued that the proposed model has implications for strategies of remedial intervention and perhaps prevention.

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BEHAVIORAL MANNERISMS OF THE BLIND INFANT AND CHILD

(A Synopsis)

June O. Bond, M.A., A.G.S.

Self-stimulatory behaviors are "repetitious, functionally maladaptive, topographically invariant motor behaviors or action sequences in which reinforcement is not specified or is noncontingent and the performance of which is regarded as pathological" (Baumeister & Forehand, 1973, p.57). These behaviors, which include but are not limited to eye-poking, body-rocking, light-gazing, and finger-flicking, are prevalent in populations of blind, autistic, retarded, and multihandicapped individuals, and are also common in normal infants. Surveys have indicated that 2/3 of institutionalized severely retarded people engage in some form of self-stimulatory behavior (Berkson & Davenport, 1962).

Originally referred to as blindisms, self-stimulatory behaviors were thought to only occur in blind individuals, and to be caused by blindness. Explanations for the appearance of these patterns in the blind date back to the early 1900's (Morse, 1965). In the 1960's, attention turned toward the presence of similar behaviors in other populations. Lovaas first used the term "self-stimulatory" to describe such behaviors of autistic children in 1967 (Repp, Deitz & Speir, 1974). In the literature "stereotypic behaviors," "stereotypies," "blindisms," "mannerisms," "autisms," and "autistic behaviors" are synonymous with "self-stimulatory behaviors," the term that is used in this presentation.

Theories behind self-stimulatory behavior in the blind, based upon a review of the literature, will be summarized as well as possible behavioral treatment approaches. An emphasis will be placed upon the importance of consideration of treatments due to the interference of self-stimulatory behaviors with learning.

SPECIFIC LEARNING DISABILITY: A COMMUNICATIVE DISORDER

Bruce K. Shapiro, M.D.

There is no universally accepted definition of Specific Learning Disability. Degree of academic impairment, intellectual potential, associated behavioral, emotional and institutional factors are continually debated. Despite this, the general concept of a child who is underachieving relative to his potential for unknown reasons is accepted.

The area of central communication disorders lacks a common ground. One group of models focuses upon the quantity of language produced and understood. Other models focus upon qualitative aspects of language function. While each of these approaches has its proponents, no classification system has been demonstrated to be effective in terms of determining etiology, prognosis or response to treatment.

As a result of the absence of accepted nosology, direct comparisons cannot be made between Specific Learning Disability (SLD) and Central Communication Disorders (CCD). However, circumstantial evidence suggests that both are points on a spectrum of communicative dysfunction. While there have been a variety of studies looking at the relationship between language function and Specific Learning Disability, they can be categorized as follows:

1. Follow up studies of language delayed children.
2. Retrospective Analyses of the Early Development of children with Specific Learnind Disability.
3. Assessment of the language functions of Learning Disabled Children.
4. Similarities of neurological function (e.g. brain structure and softsigns) and psychological test performance between SLD and CCD
5. Population studies.

Each of these approaches has major limitations, but taken as a group they suggest that 1) Children with communication disorders do have increased rates of learning dysfunction, (2) Children with learning disabilities have persistent deficits in language function, and 3) SLD and CCD cannot be distinguished by etiology or measures of neural functioning.

The relationship between SLD and CCD is of more than academic interest. Disordered communication may prove to be the earliest and most reliable marker of SLD. The treatment of SLD and CCD will become more sophisticated when subgroups can be identified and their interrelationships understood.

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Outline of Presentation

Normal and Abnormal development
of Feeding - J.F. Bosma, M.D.

Concept: feeding develops as a succession of competencies: initially pharyngeal swallow, then sucks, then oral search and enclosure of nipple. The performances of pharyngeal airway maintenance and of upper respiratory coordinations (including cry) are developing at the same time, with separate representations in the brain-stem but employing the same motor effector apparatus.

Adequate suckle feeding requires:

Arousal appropriate to the feeding effort.

Sensory elicitation appropriate for nipple enclosure, suckle and swallow. The mucosal sensory receptors and the central sensory connections are undergoing extensive development in the fetus and infant.

Central representations of the feeding actions. Development of these representations depends principally upon sensory input.

Motor apparatus which is grossly adequate. Adequate suckle and swallow can be accomplished despite most malformations of the palate, tongue or mandible, if the local sensorium is appropriate to the elicitation of the needed adaptations of motor performance.

The Relationship Between Oromotor Dysfunction and General Movement Development

Lecturer: Rona P. Alexander, Ph.D., CCC-S
Director, C.P. Project
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9001 West Watertown Plank Road
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Early detection of oral-motor problems in infants and, when necessary, the appropriate development of treatment plans and intervention strategies are dependent upon the professional's knowledge of the processes by which normal and abnormal oral movements develop as well as their relationship to the development of normal and abnormal general movement components. Whether we are talking about the development of fine-motor oral movements or general gross-motor movements, it is necessary to go beyond the traditional approach based upon developmental milestones. Rather, it requires an ability to analyze the actual components (extension, flexion, rotation) of movement development and their relationship to postural (anti-gravity) control.

As we have learned more about the development of normal movement components in regard to kinesiological, anatomical, physiological, and sensory-motor aspects, its effect on and response to oral-motor functioning becomes more evident. The development of more active, anti-gravity proximal movement provides a base for more advanced distal movements such as those of the oral mechanism used for feeding and phonation. These advances in oral functioning integrate with proximal activity and control to allow for further development of the normal infant's general movement abilities.

The correlation between general movement and oral-motor functioning in the normal developmental process suggests the existence of a similar relationship between the development of abnormal components of movement and abnormal oral-motor functioning in infants and children with cerebral palsy or other related

disorders. As the infant with neuromotor involvement attempts to move against gravity, abnormal sensory-motor feedback is produced resulting in greater, habitual abnormal movements. Since some of the earliest sensory-motor activities of the infant directly involve the use of the oral mechanism (feeding, crying, mouth-to-hand activities), oral-motor dysfunction is often an early indicator of neuromotor difficulties.

This presentation will attempt to point out significant aspects of the normal components of oral-motor and general movement development within the first year of life and their relationship to each other. Abnormal oral movements will be described and their significance discussed in regard to general movement development.

Oromotor Facilitation in Pre-Speech

Lecturer: Rona P. Alexander, Ph.D., CCC-S
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An appropriate, individualized pre-speech treatment program must be developed once an infant or child has been identified as having abnormal or significantly delayed oral-motor, respiratory/phonatory, and/or early sound production/communication activities. All aspects of the treatment program should emphasize the child's need for stimulation of more normal responses with a reduction or inhibition of abnormal responses.

When developing a pre-speech treatment program, it is necessary to recognize the differences between goals, objectives, and treatment procedures established for progress in treatment sessions and those established for carryover and maintenance during the individual's daily activities. When no differentiation is made between these two programming areas, frustration, confusion, and limited cooperation generally result.

The feeding area has often posed the greatest challenge to those developing pre-speech treatment programs. When an infant or child exhibits difficulties in feeding activities, it is often an automatic response to establish mealtime as the time when changes are to be stimulated in the child's oral-motor activity. By doing this, the realistic and necessary mealtime goal of feeding a meal with maximal nutritional intake becomes overshadowed by treatment goals that may emphasize increased active lip closure, a greater variety of active tongue movements, more actively controlled jaw stability, and greater coordination of oral movements with respiration.

Of course, there must be a constant interaction between progress in treatment and its carryover into the child's daily activities. However, it is totally

unrealistic to expect intensive work on oral-motor coordination to be restricted to the mealtime. As advances are made in treatment, they should be incorporated into the regular mealtime routine so that their effect is as positive as possible.

This presentation will more fully discuss significant aspects of pre-speech treatment programming, especially in regard to improving oral-motor functioning in feeding and early sound production. The important influences of handling, positioning, and sensory stimulation on oral-motor coordination will be emphasized.

Management of the Child with Speech Motor Dysfunction

Sally W. Blackstone, Ph.D., CCC-Sp
John F. Kennedy Institute
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Children who fail to develop speech at expected times require further evaluation by specialists in the area of developmental disabilities. Delayed speech is associated with a range of developmentally disabling conditions, many of which may be identified during the first year of life. These include cerebral palsy, cleft palate, hearing impairment, Down's syndrome, severe mental retardation and pervasive developmental disability. Other children, who are slow or deviant in their development of sound making and speech, may not be multiply handicapped. However, they will also benefit from early identification.

If a child is found to have normal hearing, and a discrepancy is felt to exist between cognitive and language abilities or between receptive and expressive language abilities, an in-depth evaluation of the child's speech motor status is indicated.

Conversational speech, produced at a rate of approximately 190 words per minute, requires the coordination of one hundred muscles. Speech-language pathologists are trained to evaluate the speech mechanism and to identify problems that interfere with speech intelligibility. These include disorders of phonation, articulation, resonance and fluency. In the older child oral motor batteries, which assess a child's non-speech and speech motor movement, articulation tests and speech intelligibility measures are used to delineate problems interfering with speech production. However, infants and younger children require other evaluation techniques. Often the clinicians' tools are limited to a parent interview, structured observation of the child and completion of a developmental checklist. Some oral motor abilities can be assessed using feeding scales since young children are generally willing to eat. However, this is not an adequate means of determining a child's speech motor status. A sample of speech is necessary, although it is difficult to obtain from a young child in a clinical setting. Thus, clinically useful procedures to elicit representative samples of pre-speech and speech behaviors in infants and young children are needed.

Since the primary function of speech is communication, a child whose neuromotor dysfunction interferes with his ability to talk is likely to be communication disordered. Therefore, the primary objectives in the management of children with speech motor dysfunction are: 1. to improve the child's speech intelligibility and 2. to improve the child's ability to communicate in all situations.

A variety of management strategies are reported in the literature with varying degrees of success. These include:

Surgical intervention, to repair structural abnormalities
Cerebellar stimulation (Robertson, Meak and Smith, 1980).
Speech prostheses (e.g., speech bulb, palatal contour) (Goyal and Greenstein, 1982).
Oral neuromotor treatment (Wilson, 1977).
Articulation therapy (Chappell, 1973; Yoss and Darley, 1974)
Assistive communication devices (Vanderheiden, 1978).

This paper is limited to a brief review of those treatment approaches in which the speech-language pathologist assumes the primary role. General treatment principles will be delineated and specific examples of treatment will be used to illustrate how objectives might be met.

General treatment principles include, but are not limited to the following:

1. Early identification and a thorough description of neuromotor speech problems is required.

2. Treatment objectives to remediate the speech motor problems should be based on a modified developmental approach.

3. Treatment procedures should utilize a learning theory paradigm within a naturalistic and communicative context.

4. The child's communication partners should be involved in the treatment program.

5. Compensatory strategies should be considered for use by the child, family and teachers.

For the child with severe speech motor dysfunction, communicative needs often outweigh their ability to speak so that others can understand them. These children may be assisted in their attempts to communicate through the use of a communication prosthesis. These may include computers, electronic devices, switches, language boards, manual sign systems, graphic symbol systems, such as Blissymbolics, speech synthesizers or a simple yes/no response system. Criteria for the development of an alternate or augmentative communication system will be discussed.

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Harvey S. Singer, M.D.
February 1982

The Neurology of Oromoter Dysfunction and Language Disorders

A neurologic examination which analyzes systems, pathways and brain regions can frequently localize sites of dysfunction. Once the anatomic lesion is defined, the appropriate specialist can then be selected to further evaluate the defective function and develop a program of remediation.

I. General Factors

The neurologic examination of a child with a language disability, begins by excluding general factors which may interfere with language development. These include mental retardation, rare cases of psychosocial deprivation, or a lack of incentive to communicate. Once general factors are eliminated, the evaluation can simplistically be separated into three phases: an evaluation of input, central processing and output.

II. Disorders of Input

Disorders of input include problems with detecting a sound stimuli, transmission through the middle ear, encoding in the inner ear, transmission along the eighth nerve and the ability to decode. In a child with adequate intellect, no evidence for CNS dysfunction and adequate stimulation, auditory disorders represent the most frequent etiology for language dysfunction. Hence, it is extremely important that hearing be adequately tested in every child with defective speech or language development.

III. Disorders Affecting Speech Output

This category contains children who have access to language symbols, but are unable to produce meaningful sounds. Indistinct speech may be produced for a variety of reasons including pseudobulbar, bulbar, cerebellar and neuromuscular defects. The neurologic examination must therefore dissect both the individual peripheral components needed to produce speech (the trigeminal, facial, glossopharyngeal, vagus, accessory and hypoglossal cranial nerves; diaphragmatic and intercostal muscles) and their ability to function in a coordinated fashion.

IV. Central Processing

The central processing of language, albeit extremely complex, will be reduced in this lecture to a simplified scheme. In this functional model, brain regions will be assigned specific language functions. Utilizing this framework, we will discuss the concept of language dominance, developmental language disabilities and aphasia.

GENETICS OF AUTISM

Joe Matthew Farber, M.D.

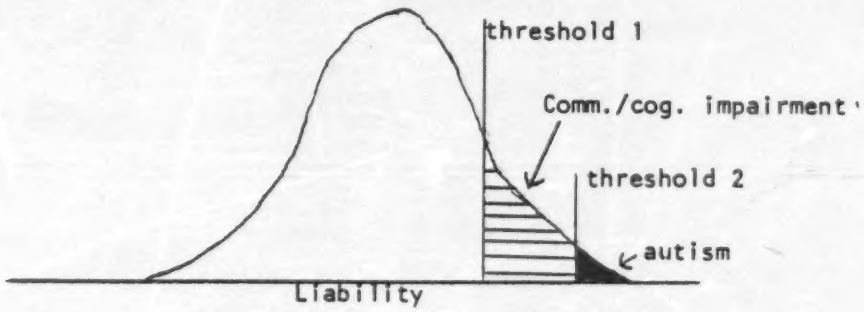
Autism is a familial condition. While data has not been published about the offspring of patients with autism, the recurrence risk for siblings is 2%, approximately 50 times that of the general population. While some cases of autism follow single gene inheritance patterns (e.g. PKU, possibly fragile X), most cases of autism do not fit the classical Mendelian constructs of autosomal recessive, autosomal dominant, or X-linked. As a consequence, a multifactorial model has been proposed.

One approach to determine the role of genetics in causing autism is by twin studies. Monozygotic (MZ) twins share genetic material fully, while dizygotic (DZ) twins have only 50% overlap. Since differences in intrauterine environment in a set of twins are minimized (but not eliminated), any differences between the two subtypes would be expected to be due to genetic causes. Folstein and Rutter studied 21 twin pairs. Of 11 MZ pairs, 4 were concordant for autism and an additional 5 for other cognitive problems; there were no DZ twins concordant, and only 1 of 10 siblings showed cognitive difficulties. This suggests a genetic etiology, although concordance was not 100%. In addition, there was evidence for environmental influence, in that brain injury was reported to account for most of the discordant cases, but not for the concordant ones.

The above study clearly indicates that both genetic and environmental factors play a role in the etiology of autism. A multifactorial model takes this into account. Such a model is based on the following assumptions:

a. Polygenetic and environmental influences combine to yield one variable, the liability;
b. Liability is distributed in a Gaussian fashion;
c. Each gene affecting liability is, individually, of little effect;
d. The population can be divided into different classes based on different thresholds.

Adapting this model to autism, liability will be on a scale of communicative/cognitive abilities (see diagram below). One threshold would be for autism proper; a combination of environmental and genetic influences exceeding the threshold results in autism. At the other end of the spectrum is normal functioning. In between, there will be another threshold creating a third class of patients, with communicative/cognitive impairment.



Such a model can be verified mathematically. Hypotheses can be generated and compared to existing data. Among predictions to be made are the following:

1) The rarer the condition, the <u>greater</u> the relative risk for first degree relations;
2) Probands at high genetic risk would require little environmental influence to develop autism; family members with similar risks but a more benign environment would be prone to cognitive difficulties;
3) Where autism is due to severe environmental insult (e.g. congenital Rubella), recurrence risks should be low;
4) More severely affected probands would have more severely affected relatives;
5) Since autism predominates in males, the recurrence rates for siblings of female probands should be <u>greater</u> than that for males.

For the first hypothesis, the more common a condition, the lower the presumed threshold, and thus the less the average genetic difference between first degree relations of probands and the general population. Because the differences between these two populations are small, the relative risk is less than with a rare condition.

For comparison, the incidence of congenital dislocation of the hip (in females) is .002, with a relative risk for first degree relations of 25. The numbers for cleft lip are .001 and 40 respectively, while for autism the incidence is .0004, and the relative risk 50, supporting the model.

The second hypothesis has recently been explored more fully. August et al., for example, found 11/71 siblings of autistic children to have cognitive problems, compared to 1/38 in a control group. Minton et al., found 50 siblings of autistic children to have an average full-scale I.Q. of 88, with the expected average being 98. Both of these studies are consistent with the multifactorial model. For the remaining 3 hypotheses, although the necessary data should be obtainable, they are not yet available. The final answer to the question awaits further data.

Jon Matthew Farber, M.D.

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Metabolic Approaches to Autism and Related Disorders

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While Kanner formulated a psychological origin for infantile autism (1) current thinking favors a biological basis. Why is this the case? In part it relates to the demonstration of genetic factors, most strikingly the finding of 50% concordance in identical twins (2,3). Perhaps, it is also part of a current trend toward emphasis of biological factors in psychiatry, and our increasing awareness that abnormalities in transmitter function or lesions in specific brain areas could indeed lead to disturbances of function and behavior that are characteristic of autism. While such analogies and general concepts clearly are of value for the formulation and testing of hypotheses, the question I would like to consider briefly today is to what extent such specific biological factors have indeed been identified in autism: In this discussion, we will not consider autism-like symptoms that may be associated with entities such as phenylketonuria, viral disorders, or the fragile X-syndrome. The main point of interest is to inquire about the nature of autism which is not associated with defined etiological entities. Is "idiopathic" autism a single entity, can it be related to any known metabolic disturbance? This issue has been critically reviewed in a recent publication (4).

Before reviewing the data on biochemical abnormalities, it is important to determine if there is a demonstrable structural abnormality of the nervous system in autism. It was clear long ago that patients with "idiopathic" autism did not have major structural abnormalities recognizable by classical neuropathological techniques. However, it is widely recognized that these

older techniques might not detect more subtle changes in synaptic connections, or dendritic arborizations. An important contribution to this question is a recent authoritative study utilizing modern techniques, which failed to demonstrate structural abnormalities (5). This study focused on persons who were severely involved and functioned in the mentally retarded range. The fact that even here no structural abnormalities were found makes it unlikely that changes would be demonstrable in more mildly involved persons.

How about biochemical changes? The very extensive literature on this topic is reviewed in the previously cited reference (4). One important observation is that 40% of autistic persons have elevated levels of blood serotonin (6). This observation led to a recent study which has attracted a great deal of interest. In this study three autistic boys with elevated blood serotonin levels received fenfluramine. One of these autistic boys had an identical twin who was similarly involved. This medication, which is sometimes used as an anorexigenic drug, is known to lower brain and blood serotonin levels. The medication was given in a double blind, crossover study. It brought about a lowering of blood serotonin levels as well as some improvement in psychological functioning. The significance of the result was heightened by the fact that there was correlation between biochemical and clinical changes, and that during this period no changes were observed in the concordant and untreated identical twin of one of the patients. Implications of this study are discussed in an editorial (7), and possible mechanisms by which alterations in serotonin could affect autistic behavior are discussed in a recent review (8). The authors emphasize that these are preliminary studies with only a limited number of patients, and selected to include those who had high blood serotonin levels. It is also important to emphasize that 90% of serotonin in the human body is located in the enterochromaffin cells of the

gastrointestinal tract and thus not directly related to the central nervous system. It is not known whether the fenfluramine effect on serotonin involved the visceral or nervous system compartments or both. It should also be noted that hyperserotonemia is observed in conditions other than autism. Favorable effects on psychological function in autism have also been reported with haloperidol (9) a neuroleptic which affects dopamine metabolism, and thus has a mechanism of action which differs from fenfluramine.

In conclusion, while there are some intriguing leads about possible biochemical abnormalities and pharamcological therapeutic approaches to autism, these are not yet conclusively established. Recent advances in neurochemistry and neurobiology, and the development of new non-invasive techniques of study provide hope that meaningful new insights may be achieved in the relatively near future.

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